Br Heart J 1985; 54: 583–92

Congenital heart malformations in the first year of life—a necropsy study

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SUMMARY The hearts of 291 babies with congenital heart disease who died before the age of one were examined systematically by sequential segmental analysis to determine the lesions that were present. There was an abnormal connection between the cardiac segments in one third of cases. Patient ductus arteriosus, which is usually a common defect, was not an important finding at necropsy, whereas common arterial trunk (a rare defect) was found in 10%. The clinical and necropsy incidence of complete transposition was similar to that reported in earlier studies. The incidence of some lesions present at necropsy (for example Fallot's tetralogy) has altered over the past 10 years. A decline in the frequency with which a lesion is detected at necropsy may indicate advances in treatment or differences in classification. The presence of additional lesions influences the prognosis and subcategorisation within the major defect groupings gave some insight into the cause of death in many of the cases.

In the West congenital heart disease is the most common cardiac condition of childhood. It may affect as many as 2% of livebirths[1 2] and many babies with congenital heart disease die within the first few weeks of life. More die, with or without surgical intervention, before the end of their first year.

More specific classification of these cardiac lesions depends on a systematic approach to the diagnosis of congenital heart malformations, such as that proposed by Van Praagh.[3] With some modifications[4] we have used this approach to analyse a large series of necropsy cases aged one year and younger collected over the past ten years at this hospital.

Patients and methods

We studied 291 hearts from patients who died within the first year of life. These hearts comprised 72% of the 403 specimens that were collected by the Paediatric Cardiac Morphology Laboratory, Cardiothoracic Institute, University of London from October 1974 to October 1984. Most came from patients attending the Brompton Hospital, while a small proportion were referred from other centres in the United Kingdom.

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Accepted for publication 10 September 1985

The specimens were divided into two age groups—those aged < 4 weeks and those aged from 4 weeks to one year.

We systematically examined each heart by a sequential segmental approach[4] and noted any abnormal connections or arrangements of the cardiac segments. We then ascribed a primary diagnosis to each heart. Hearts which showed an absent left atrioventricular connection, with a main chamber of right ventricular morphology (classical mitral atresia) and hearts with an imperforate mitral valve were grouped with specimens having aortic atresia. This was so that our series could be compared with clinical series in which these conditions were combined. In hearts with multiple lesions the primary lesion was the one regarded as being of greatest morphological importance. In some cases these primary lesions were further categorised by subgroup.

The incidence of each lesion in both age groups was calculated as a percentage. This necropsy incidence was compared with the clinical incidence at the Brompton Hospital,[5] and both these sets of data were then compared with data from other centres.[6 7]

Results

Table 1 shows the sequential segmental arrangement and connections in the hearts that we examined. Two of the four specimens with left atrial

Table 1 *Sequential segmental arrangement and connections of hearts from 291 cases < 1 year*

	No	%
Atrial arrangement:		
Normal (situs solitus)	276	95·2
Right atrial isomerism	11	3·8
Left atrial isomerism	4	1·4
Atrioventricular connection:		
Concordant	246	84·5
Discordant	1	0·3
Biventricular ambiguous connection		
Left hand topology (l loop)	3	1·0
Right hand toplogy (d loop)	4	1·4
Double inlet		
Right ventricle	4	1·4
Left ventricle	4	1·4
Indeterminate ventricle	4	1·4
Absent right connection		
Dominant right ventricle	1	0·3
Dominant left ventricle	6	2·1
Indeterminate ventricle	1	0·3
Absent left connection		
Dominant right ventricle	16	5·5
Dominant left ventricle	1	0·3
Ventriculoarterial connection:		
Concordant	174	59·8
Discordant	32	11·0
Double outlet		
Right ventricle	28	9·6
Left ventricle	4	1·4
Indeterminate ventricle	4	1·4
Single outlet		
Common arterial trunk	23	7·9
Pulmonary trunk	9	3·1
Aortic trunk	37	12·7

isomerism showed an ambiguous biventricular atrioventricular connection. Each case was connected to a ventricular mass having right hand topology (d loop). Five of the 11 cases of right atrial isomerism had ambiguous connections. Two cases had right hand ventricular topology and three had left hand topology (l loop). There was a univentricular atrioventricular connection in 37 specimens. There were 17 examples of absent left atrioventricular connection, of which 16 cases were classic mitral atresia. In each of these, the right atrium was connected to a dominant right ventricle. In the remaining case the right atrium drained to a dominant left ventricle.

Sixty per cent of the cases had concordant ventriculoarterial connections. There were discordant connections in 32 (11%) hearts. There were 36 (12·4%) specimens with double outlet connection. The remaining 69 (23·7%) cases had single outlet connections.

Thirteen primary lesions were identified; each accounted for more than 4% of the necropsy cases (Table 2). Each group was subdivided to give a more comprehensive view of the spectrum of malformations (Table 3). Data on surgical experience are also included. The most frequent condition found at necropsy in the first month of life was common arterial trunk. Fifteen (9·9%) of the 151 hearts had this lesion (Table 3). Of the 15, five cases had the additional lesion of interruption of the aortic arch and a further two cases had other arch anomalies. Another two cases showed truncal valve incompetence. Total correction had been attempted in one case with interruption and in two other cases. This lesion also showed an appreciable incidence in the older group, accounting for 5·7% of hearts from those aged between one month and one year. Of the eight cases, three had truncal valve stenosis and one of these also had major accessory systemic-pulmonary collateral arteries. Another case had partial anomalous pulmonary venous connection and a fifth had arch hypoplasia. Four cases had undergone complete repair, after which one case developed atrioventricular heart block. A palliative operation had been performed on one heart in each age group; in the younger case the heart had additional interruption of the aortic arch and the older case had severe truncal valve stenosis.

In the younger age group the incidence of mitral and aortic atresia or both (9·9%) was considerable. There were 11 cases of mitral atresia; five had additional aortic stenosis and isthmal hypoplasia, and five had aortic atresia. Aortic atresia alone was seen in four cases all of which had mitral valve stenosis. Isolated aortic atresia was not seen in the older group and there were only five cases of mitral atresia—two with additional pulmonary atresia, one with double outlet right ventricle, and two with hypoplasia of the aortic arch. Three cases from each age group had had palliation.

Hearts with a univentricular atrioventricular connection (excluding classic mitral atresia)—that is those with no right/left atrioventricular connection and a double inlet ventricle—comprised 9·3% of the hearts of those who died during the first month of life. There was one case of classic tricuspid atresia, two cases of tricuspid atresia with complete transposition (both cases having aortic obstruction and isthmal hypoplasia), one case of absent right atrioventricular connection to an indeterminate ventricle with pulmonary atresia, one of absent right atrioventricular connection with the left atrium connecting to a dominant right ventricle; one of absent left atrioventricular connection with the right atrium connecting to a dominant left ventricle; three cases of double inlet left ventricle (two with aortic isthmal hypoplasia and double outlet left ventricle, the other with pulmonary atresia and right ventricular origin of the aorta); three cases of double inlet right ventricle (one with pulmonary atresia and two with atrial isomerism and its usual accompanying lesions), and two cases of double inlet to an indeterminate ventricle (both with atrial isomerism). Palliative procedures had been performed in six hearts. In addi-

Table 2 *Necropsy incidence of the primary lesions found in the two age groups*

	Incidence (%) newborn–1 mth (n=151)	*Incidence (%) 1 mth–1 yr (n=140)*
Common arterial trunk	9·9	5·7
Arch interruption	8·6	1·4
Arch hypoplasia including discrete coarctation	9·3	7·1
Univentricular atrioventricular connection*	9·3	5·0
Complete transposition	7·3	10·0
Mitral atresia and aortic atresia or both	9·9	3·6
Pulmonary atresia with intact septum	7·3	7·2
Double outlet right ventricle	4·0	8·6
Anomalous pulmonary venous connection	7·3	7·2
Atrioventricular septal defect	3·3	15·0
Ventricular septal defect	4·6	5·0
Pulmonary atresia with ventricular septal defect	1·3	3·6
Fallot's tetralogy	0·6	4·3

*Excluding classic mitral atresia.

tion, 5% of hearts from the older group had univentricular atrioventricular connection. Of the seven hearts, only one case—with double inlet left ventricle—showed atrial isomerism. Pulmonary outflow tract obstruction was seen in two of three cases of classic tricuspid atresia. There were two cases of double inlet to a solitary and indeterminate ventricle; one of these also had double outlet from the solitary ventricle and complete correction had been attempted. There was one example of double inlet right ventricle. Four cases had had palliative surgery.

The incidence of hypoplasia of the aortic arch in the younger group (9·3%) resembled that of univentricular atrioventricular connection. There were 14 cases in total, eight of which had associated ventricular septal defects. Discrete coarctation occurred in four cases, coarctation with tubular hypoplasia in two, and tubular hypoplasia alone in eight cases. Six of the cases had aortic outflow tract obstruction (Fig. 1a) and a further two cases had a bicuspid aortic valve. The incidence of arch hypoplasia was also high (7·1%) in the older age group. Six of the 10 hearts had discrete coarctation. Of these, two had left ventricular outflow tract obstruction, one had a bicuspid aortic valve, one a persistent left superior caval vein, and one had endocardial fibroelastosis. The remaining four hearts with tubular hypoplasia all had outflow tract obstruction and a ventricular septal defect.

Thirteen cases (8·6%) of interrupted aortic arch were to be seen in the younger age group. All but one case also had a ventricular septal defect. Eight cases had some obstruction to the subaortic outflow tract (Fig. 1b) and seven hearts had undergone total correction of the malformation. In contrast, arch interruption occurred only twice (1·4%) in the 140 cases aged between one month and one year. One case had a subaortic ventricular septal defect and double outlet left ventricle and the other had an aortopulmonary window. In both cases surgical correction had been attempted.

The incidence of pulmonary atresia with intact septum and anomalous pulmonary venous connection was 7% in both age groups. Four of the eleven younger cases of pulmonary atresia with intact septum had Ebstein's malformation of the tricuspid valve. Three of the eleven had dilated, thin-walled right ventricles (Fig. 2a), while the remaining eight had small ventricular cavities resulting from gross trabecular hypertrophy or ventricular hypoplasia (Fig. 2b) Of the 10 cases in the older group, two had dysplastic and incompetent tricuspid valves and eight had right ventricular cavities which were obliterated by trabecular hypertrophy. All had been treated by palliative procedures. There were eleven cases of anomalous pulmonary venous connection in the group less than one month old. Five cases had an infradiaphragmatic anomalous connection. In contrast, none of the 10 cases aged between one month and one year showed an infradiaphragmatic connection.

Complete transposition was also important in the younger and older age groups, with an incidence of 7·3% and 10·0% respectively. Three of the eleven postnatal cases had associated coarctation or isthmal hypoplasia. In all, palliative surgery was performed on nine cases and total correction had been attempted on twelve.

The incidence of atrioventricular septal defect was fairly low (3%) in the younger age group. It was the most important of all the lesions in the older group (15%), however. Twenty two of the 26 cases had associated major complicating lesions—severe right ventricular dominance in three (Fig. 3); double outlet right ventricle in three; arch hypoplasia in six; pulmonary atresia in two; and left ventricular outflow tract obstruction in four. Surgery had been

Table 3 *Classification and subclassification of lesions found at necropsy and the operations performed in these cases*

Lesion	*< 1 mth*			*1 mth–1 yr*			*Associated conditions*
	No	*Surgery*	*Palliation*	*No*	*Surgery*	*Palliation*	
Common arterial trunk	10	2	—	8	4	1	Valve incompetence stenosis (5)
+ interruption	5	1	1	0	—	—	
Arch interruption	1	—	—	1	1	—	AP window (1)
+ VSD	4	3	—	0	—	—	
+ VSD and LVOTO	8	4	—	1	1	—	DOLV (1)
Coarctation	1	1	—	3	3	—	AVSD (1)
+ LVOTO	2	1	—	2	2	—	Arch hypoplasia (1)
+ VSD	3	1	—	1	1	—	Arch hypoplasia (1), ASD (1), VA discordance and RVOTO (2)
Arch hypoplasia	3	2	—	0	—	—	
+ VSD	5	2	—	4	4	—	LVOTO (7)
Ventricular septal defect	5	—	2	3	1	2	Mild arch hypoplasia (6); multiple defects (2)
+ LVOTO	2	—	1	2	2	—	
+ RVOTO	0	—	—	2	2	—	
Mitral atresia	4	—	—	2	—	1	
+ aortic atresia	5	—	1	0	—	—	Aortic stenosis (5)
+ pulmonary atresia	0	—	—	2	—	1	
+ DORV	2	—	2	1	—	1	Aortic stenosis (2)
Aortic atresia with mitral stenosis	4	—	—	0	—		
Pulmonary atresia							
+ dilated RV	3	—	—	2	—	2	Ebstein's (2), incompetent tricuspid valve (3)
+ small RV	8	—	3	8	—	8	Ebstein's (2), incompetent tricuspid valve (2)
Double outlet RV	6	1	3	9	2	2	Arch hypoplasia (5)
+ RVOTO	0	—	—	3	3	—	
Anomalous PV connection							
Infradiaphragmatic	5	4	1	0	—	—	
Supradiaphragmatic	5	2	—	7	7	—	
Unknown	1	—	—	3	—	1	
Univentricular AV connection							
Absent right	5	—	4	3	—	2	TGA (3), DOLV (1), RVOTO (2), pulmonary atresia (1)
Absent left	1	—	—	0	—	—	
Double inlet LV	2	—	1	0	—	—	DOLV (2)
+ pulmonary atresia	1	—	—	1	—	1	Right isomerism (1)
Double inlet RV	0	—	—	1	—	—	DORV (1)
+ RVOTO	3	—	1	0	—	—	Pulmonary atresia (2), DORV (1)
Double inlet indeterminate ventricle	2	—	—	2	1	1	Right isomerism (2)
Atrioventricular septal defect	2	—	—	15	10	—	Arch hypoplasia (6), RV dominance (3), mitral stenosis (2), hypoplastic RV (1), anomalous PV connection (1)
+ LVOTO	3	—	—	1	1	—	TGA (1)
+ pulmonary atresia	1	—	1	1	—	—	
+ DORV	0	—	—	3	2	—	
Pulmonary atresia + VSD							
+ ductal supply	2	—	2	4	—	2	Aortic override (2)
+ MAPCAs	0	—	—	1	—	1	
Tetralogy of Fallot	1	—	1	6	5	1	Aberrant PA (2), DORV (1)
Complete transposition	11	3	5	14	9	4	Mild arch hypoplasia (4)

AP, aortopulmonary; ASD, atrial septal defect; AVSD, atrioventricular septal defect; DOLV, double outlet left ventricle; DORV, double outlet right ventricle; LV, left ventricle; LVOTO, left ventricular outflow tract obstruction; MAPCAs, major accessory pulmonary collateral arteries; PA, pulmonary artery; PV, pulmonary venous; RV, right ventricle; RVOTO, right ventricular outflow tract obstruction; TGA, transposition of the great arteries; VA, ventriculoarterial; VSD, ventricular septal defect.

performed mainly on the older group; 13 of the 21 having had attempted total correction.

Double outlet right ventricle was found in 4% of the younger group and 8·6% of the older group. Three of the six younger hearts also had arch hypoplasia; only one had been repaired. The remaining three cases had had palliative procedures. Of the twelve cases in the older group, six had subpulmonary ventricular septal defects. Of these three had severe right ventricular subaortic outflow tract obstruction and underwent attempted correction. A further two cases had surgical correction and palliation was performed on another two.

Fallot's tetralogy was not an important finding in the younger group. There were six cases in the age group one month to one year. Two of these had additional malformations of the aortic arch. Another had a narrow pulmonary trunk and double outlet

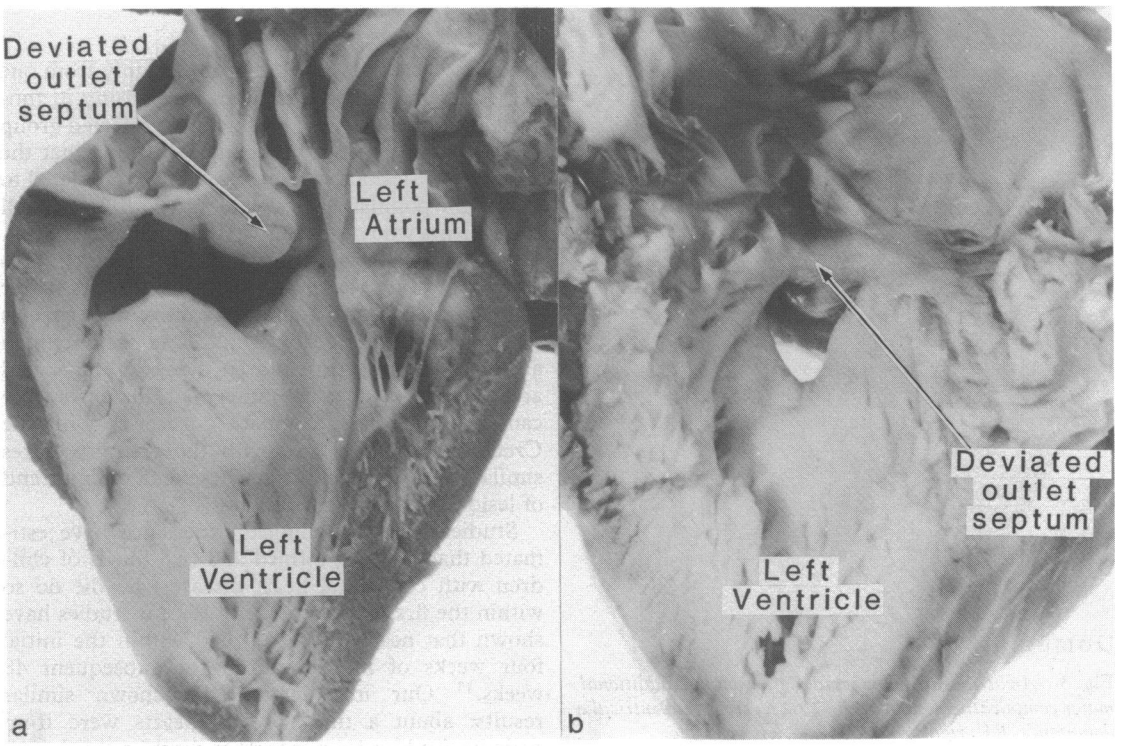

Fig. 1 *Malalignment ventricular septal defect resulting in subaortic outflow tract obstruction in (a) aortic arch hypoplasia and (b) interrupted aortic arch.*

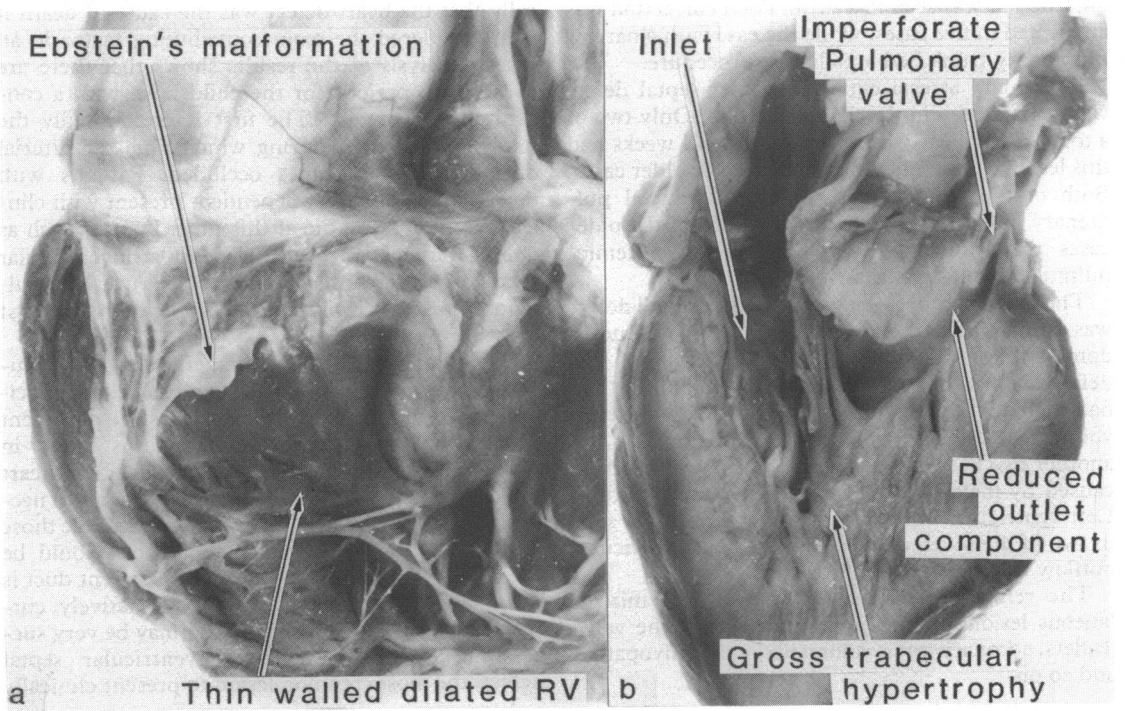

Fig. 2 *The two morphological patterns of pulmonary atresia with intact septum. (a) Dilated thin walled right ventricle (RV) associated with Ebstein's malformation of the tricuspid valve. (b) Gross trabecular hypertrophy causing virtual obliteration of the apical trabecular and outlet components.*

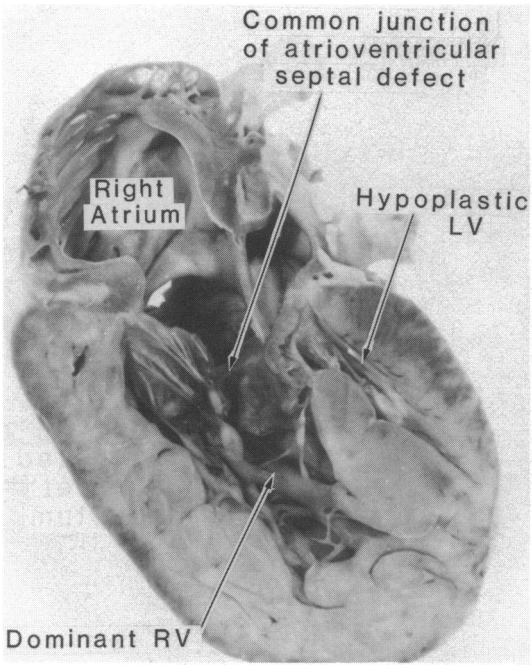

Fig. 3 *An atrioventricular septal defect with an additional major complicating lesion, in this case severe right ventricular dominance. RV, right ventricle; LV, left ventricle.*

right ventricle, a fourth had a bicuspid pulmonary valve, and severe right ventricular outflow tract obstruction was seen in a fifth. Total correction was attempted in five cases, while the case with a narrow pulmonary trunk had a palliative procedure.

Pulmonary atresia with ventricular septal defect was seldom seen in the younger group. Only two of a total of 151 hearts from those aged <4 weeks had this lesion compared with five of the 140 older cases. Both of the younger cases showed a ductal pulmonary blood supply while at least one of the older cases was known to have had major systemic-pulmonary collateral arteries.

The frequency of isolated ventricular septal defect was similar in both groups although it was not a dominant lesion in either. Of the 14 hearts with this defect five had had repair of the defect and five had had palliative procedures. In four of the seven younger cases there was mild arch hypoplasia and another two had left ventricular outflow obstruction caused by malalignment of the ventricular septum. Left ventricular outflow tract obstruction was seen in two of the seven older cases and right ventricular outflow tract obstruction in a further two.

The remainder of the 291 hearts had miscellaneous lesions such as malformations of the valve leaflets, coronary artery anomalies, cardiomyopathy, and so on.

Discussion

Necropsy cases in this study came mainly from one centre in the United Kingdom specialising in thoracic medicine. As such, it is a highly selected group of cases and it would be wrong to assume that the incidence of lesions seen here can be extrapolated to that in the population as a whole. In addition, not all the patients who die in our centre come to necropsy. It is thus impossible to calculate mortality figures from clinical and necropsy incidence data. A better estimate of this can be obtained from studies such as that recently reported by Samanek *et al*,[8] in which all the stillbirths and children dying in a specified area of Czechoslovakia underwent necropsy. We can, none the less, compare our series with the Czech investigation and with those from centres similar to our own and from these data infer a trend of lesion incidence and prognosis.

Studies based on clinical experience have estimated that between a third and two thirds of children with congenital heart disease who die do so within the first year of life.[1910] Recent studies have shown that nearly as many died within the initial four weeks of life as did in the subsequent 48 weeks.[11] Our investigation has shown similar results: about a third of our hearts were from patients aged under a month at death, a third were under a year, and the remainder were from children aged over a year. Although we cannot say categorically that the heart defect was the cause of death it was considered the main contributing factor in all cases. Analysis of the results shows that there are two critical periods for the child born with a congenital heart defect. The first is undoubtedly the first month of life, during which time the arterial duct normally becomes occluded. Patients with defects which are duct dependent present with clinical symptoms and die at this time. Defects such as double outlet right ventricle and atrioventricular septal defect, however, do not present until the pulmonary vascular resistance falls at the end of the first month.

Some interesting points emerge from a comparison between necropsy data and the clinical incidence at our centre (Table 4). Firstly, persistent patency of the arterial duct (which is found in 10–11% in children born with congenital heart disease[1 12]) had a clinical incidence of 6% and a necropsy incidence of $<1\%$. The results resemble those found at other centres (Table 5). They could be taken to indicate that an anatomically patent duct is not a particularly lethal lesion. Or alternatively, current management of this condition may be very successful. Secondly, "isolated" ventricular septal defect, the most common lesion to present clinically

Table 4 *A comparison of the clinical incidence (Brompton Hospital) and the necropsy incidence in a series of hearts from patients < one year old*

Lesion	*Clinical incidence 1973–82 (n=1653)*	*Necropsy incidence 1974–84 (n=291)*
Ventricular septal defect	15·4	4·8
Complete transposition	10·9	8·6
Coarctation and hypoplasia	10·5	8·3
Tetralogy of Fallot	9·9	2·4
Persistent arterial duct	6·3	0·6
Atrioventricular septal defect	3·9	8·9
Pulmonary stenosis	3·0	—
Pulmonary atresia with intact septum	1·9	7·2
Anomalous pulmonary venous connection	3·6	7·2
Tricuspid atresia	4·7	7·2*
Univentricular atrioventricular connection	4·3	
Mitral atresia and/or aortic atresia	—	6·9
Double outlet right ventricle	3·0	6·2
Common arterial trunk	2·1	7·9
Atrial septal defect	0·5	—
Arch interruption	—	5·2

*Excluding classic mitral atresia.

Table 5 *A comparison of clinical data reported by four centres*

Lesion	*Clinical incidence Brompton 1973–82 (n=1653)*	*Diagnostic frequency NERICP[6] 1969—74 (n=2381)*	*Keith et al[7] 1967 (n=1380)*	*Lesion incidence Bohemian study 1952–79 (n=2257)*
Ventricular septal defect	15·4	15·7	26·0	22·6
Complete transposition	10·9	9·9	8·8	5·3
Fallot's tetralogy	9·9	8·9	6·8	—
Coarctation	10·5	7·5	4·1	5·2
Hypoplastic left heart	3·7	7·4	2·9	4·7
Persistent arterial duct	6·3	6·1	7·2	10·2
Atrioventricular septal defect	3·9	5·0	4·2	4·2
Pulmonary stenosis	3·0	3·3	—	4·7
Pulmonary atresia with intact septum	1·9	3·1	0·7	2·6
Atrial septal defect	0·5	2·9	3·3	3·6
Tricuspid atresia	4·7	2·6	3·2	4·1
Univentricular atrioventricular connection	4·3	2·4		
Aortic stenosis	1·1	1·9	13·0*	2·4
Double outlet right ventricle	3·0	1·5	0·3	2·7
Common arterial trunk	2·1	1·4	1·8	1·7
Anomalous pulmonary venous connection	3·6	2·6	1·2	1·6

*Includes aortic atresia and mitral valve stenosis and atresia.
NERICP, New England Regional Infant Cardiac Program.

(with an incidence of > 15%) and in the Czech necropsy series (an incidence of 22%[8]) was found in only 5% of our necropsy cases. Moreover, the incidence was 5% in both age groups, whereas more than 80% of the clinical cases do not present until the pulmonary vascular resistance falls. This probably reflects the current policy of treating infants at local cardiac centres, with only serious cases being referred to national centres. The referral of serious cases to our hospital could account for the higher frequency of this lesion in the younger group and for the increased incidence at necropsy when compared with the New England study (Table 6). Thirdly, common arterial trunk, double outlet right ventricle, total anomalous pulmonary venous connection, and pulmonary atresia with intact septum—all of which have a clinical incidence of less than 5%—were more commonly seen at necropsy. Fourthly, arch malformations, complete transposition, and univentricular atrioventricular connection (which have a low natural incidence) are the most frequently referred complex congenital heart malformations and are also dominant in the necropsy material. The fact that they remain dominant despite surgical intervention in more than half of the older patients under one year is disturbing. Fifthly, the clinical incidence of aortic stenosis and pulmonary stenosis is low and these conditions are not found at necropsy. Finally, we found that one third of our cases either had a major abnormality in the arrangement of the major cardiac segments[3,4] or abnormal connections at at least one of the junctions.

Table 6 *A comparison of data from three necropsy series*

Lesion	*Brompton 1974–84 (n=291)*	*NERICP series 1969–74 (n=900)*	*Bohemian study 1952–79 (n=1090)*
Univentricular atrioventricular connection	7·2*	5·7	4·7
Complete transposition	8·6	10·3	9·8
Arch interruption	5·2	2·1	0·6
Arch hypoplasia including coarctation	8·3	8·3	6·6
Common arterial trunk	7·9	2·5	3·2
Anomalous pulmonary venous connection	7·2	5·0	3·0
Aortic atresia and mitral atresia or both	6·9	15·1	2·9
Pulmonary atresia with intact septum	7·2	6·4	4·9
Pulmonary atresia with ventricular septal defect	2·4	2·2	
Ventricular septal defect	4·8	2·9	21·9
Atrioventricular septal defect	8·9	5·2	6·4
Double outlet right ventricle	6·2	1·5	4·7
Fallot's tetralogy	2·4	4·5	7·3
Atrial septal defect	—	1·4	4·9
Persistent arterial duct	0·6	—	3·2

*Excluding mitral atresia.
NERICP, New England Regional Infant Cardiac Program.

Analysis of our data by age group (Table 2) produces different results. Atrioventricular septal defect was the most frequent lesion at necropsy (8·9%) and it was also the dominant lesion (15%) in the older age group. Its incidence in the group aged less than one month was low (3%). Nearly all of the 27 cases of atrioventricular defect were associated with major complicating lesions, including severe right ventricular dominance, double outlet right ventricle, and arch hypoplasia; this may account for the high mortality.

The necropsy incidence of arch malformations which impede aortic flow (interrupted arch and arch hypoplasia and coarctation) was 13·8%. Thirteen of the fifteen cases of interruption were less than a month old. Tubular hypoplasia was also more common in the younger group, and eight of the 12 cases with this lesion were less than one month old. Arch lesions themselves are technically amenable to surgical reconstruction but 22 of our 39 cases also had some form of subaortic outflow tract obstruction. The incidence of aortic atresia or mitral atresia or both in the necropsy series was high (9·9%) in those aged <4 weeks. This accords with clinical data which show that 82% of all cases of hypoplastic left heart syndrome present before the end of the first week of life.

The necropsy incidence of pulmonary atresia with intact septum was about 7% in both age groups. The clinical incidence is low (1·9%), both in the first month and overall. Although 60% of all cases present by day 7, the prognosis is poor as shown by the high necropsy incidence. Further investigation demonstrated that 16 of the 21 cases had tiny right ventricular cavities. Right ventricular cavity size and morphology are important,[13 14] and it was no surprise to find that most of our cases had severe hypertrophy of the ventricular wall with almost total obliteration of the apical trabecular and outlet components. These are known to be the cases with the worst surgical prognosis. Also of note was the finding of three cases with grossly dilated right ventricular cavities and thinning of the ventricular myocardium. These were all associated with severe Ebstein's anomaly of the tricuspid valve. Clinically they present with high cardiothoracic index on the chest radiograph. These patients also have a poor prognosis. It is difficult to see from the specimens what potential there is for surgical repair. In our opinion, these cases should be considered as a special subgroup of cases with pulmonary atresia and intact septum.

The incidences of anomalous pulmonary venous connection and of pulmonary atresia with intact septum are similar. Five of the eleven younger cases were known to have had an infradiaphragmatic anomalous connection. Such an anomalous connection was not found in any of the older cases. The early obstruction to the pulmonary venous return at the level of the diaphragm (or through the liver sinusoids when the venous duct closes) almost certainly explains this finding. The overall mortality rate among those cases which present during the first year of life is known to be high.[15 16] This is in part due to the moribund condition of the child on admission.[17] The poor surgical results reflect this. The success of treatment is dependent on an early diagnosis. Immediate referral to operation after cross sectional echocardiography has recently improved the results, even in cases with infradiaphragmatic connection.[18]

Both double outlet right ventricle and complete transposition showed a higher necropsy incidence in the older group (8·6% and 10·0%) than during the

postnatal period (4·0% and 7·3% respectively). Three of the 11 patients with complete transposition who died soon after birth had additional arch hypoplasia. Pulmonary stenosis is well recognised in association with complete transposition, occurring in 15%–33% of all cases.[19] Right ventricular obstruction, however, is less well documented. The observation of additional arch hypoplasia should always suggest the possibility of subaortic stenosis.[20 21]

In the necropsy series tetralogy of Fallot and pulmonary atresia with ventricular septal defect were found almost exclusively in the older group. The clinical series shows that 75% of cases do not present until after the age of four weeks. The reasons for this are unclear. The right to left shunt may increase because of pulmonary stenosis or decreasing systemic vascular resistance. Alternatively, it may be because of increasing oxygen demand. Serious cases (13·4%), however, do present within the first week of life. The absence at necropsy of cases aged less than 4 weeks suggests that management of such patients is good. Without doubt, surgical interventions influence the type of case that presents at necropsy. Since we lack the data on the number of cases that come to operation, it is not possible to say whether this is generally successful. Of note is the finding that over 30% of all the necropsy cases had undergone complete repair and a further 20% had palliative procedures. The operation may have hastened death in some cases. Since most of the cases had severe anomalies complicated by additional lesions (Table 3), however, we believe that in these cases operation was attempted on patients whose prognosis was poor from the outset.

Comparison of our data with other series shows a constant emphasis on complete transposition both in the clinical (Table 5) and necropsy (Table 6) series. This is not the case for hypoplastic left heart syndrome (aortic atresia or mitral atresia or both) for which a necropsy incidence of the NERICP series was 15% compared with our value of 7·0%. Nor is it so for interruption of the aortic arch and arch hypoplasia/coarctation. The increased clinical incidence of these has been attributed to the improvements in recognition and referral,[5] and this would also influence the necropsy incidence. Earlier diagnosis and referral would reduce the number of deaths at local centres and increase the numbers at necropsy at specialised units. There is a noticeable delay in the diagnosis of hypoplastic left heart syndrome in the NERICP series compared with our own.[5] This, along with the real decrease in incidence at the clinical level, could account for the lower incidence at necropsy seen in our series.

Our incidence of univentricular atrioventricular connection, in which we have combined hearts with double inlet ventricle with those having the absent connection variant of atrioventricular valve atresia,[22] far exceeds that found by others both clinically and at necropsy. This almost certainly relates to differences in classification.[22] In this respect, atrial isomerism was not thought of as a physiologically important defect and was, therefore, not given a group of its own. Isomerism is often associated with univentricular atrioventricular connection and under such circumstances would be classified by us as the latter. In the NERICP series hearts with both defects were classified under the heading of heterotaxia. It could be argued that associated lesions are of more haemodynamic importance than the common mixing within the ventricular mass, but the connection to us seems the most striking anatomical finding. None the less, the lack of univentricular atrioventricular connection (or its alternative appellations) in Keith's series[1] must surely be due to the classification of such hearts by their additional lesions.

The criteria used to establish the primary diagnosis in multiple defects are open to differing interpretation and this is one reason for non-uniform categorisation. To ensure that modern comprehensive studies are comparable a detailed classification is needed. Sequential analysis is the first step to comparative studies because anomalies of connections may then be determined without ambiguity. It is insufficient to establish the anatomical cause of death. For this an insight into the spectrum of complicating lesions is also required and hence some degree of subcategorisation is appropriate.

We are indebted to all who have referred their specimens to our collection. We thank staff of the Departments of Pathology and Paediatrics, Brompton Hospital, for their continuing co-operation.

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